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A significant cause of constipation and growth retardation: hirschsprung's disease

Growth retardation and hirschsprung's disease

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Abstract

Aim: In this study we aimed to draw attention to HD which may lead to mortal complications if early diagnosis and treatment are not provided for infants with gastrointestinal symptoms. Material and Method: In Mustafa Kemal University, Faculty of Medicine, Department of Pediatrics between September 2016 and March 2018 the data of the patients who were diagnosed with Hirschsprung's disease were retrospectively analyzed. In this study age, gender, percentile values of growth and development (height, weight, head circumference), neurodevelopment stages (ability to tonic neck reflex, ability to sit supported and unsupported ability to sit), first meconium time, the most common complaints and physical examination findings were examined. Results: The mean age of the 19 patients was 7.42 months (48 days-40 months). Sixteen (%84) of the patients were male and 3 (%16) of the patients were female. Growth and development values were detected in 8 of the patients in the range of 10-25, 6 of the patients in the 3-10 percentile range and 5 of the patients were under the 3rd percentile. The first meconium excretion time of the patients was an average of 3 days (49 hours-5 days). Discussion: It should be kept in mind that organic causes such as HD should be considered in children with history of delayed meconium passage in the anamnesis and if complaints have been present before the age of one year and in whom growth retardation is detected in their examinations. In this way, late diagnosis, complications, unnecessary and costly tests and treatments can be prevented in the HD.

Keywords

Constipation; Growth Retardation; Misdiagnosis

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Introduction

Hirschsprung's Disease (HD) was first described by the Indian surgeon Shushruta Samheta in the 17th century. In 1887, Harald Hirshsprung, a pediatrician from Copenhagen, described the disease in two patients and the disease was named after his own name [1-3]. HD is a motility disorder characterized by the absence of peristaltic activity in the segment of the affected intestine as a result of congenital and complete absence of the nerve plexuses of the gastrointestinal tract (myenteric: Auerbach's and submucosal: Meisner's), it occurs from a disorder in the migration of the parasympathetic ganglion cells to distal intestine, during fetal life [4-7]. In about 80% of children, the affected intestinal segment is in the rectum or rectosigmoid colon while in 10% of HD, the affected intestinal segment is in the more proximal regions of the colon. In addition, 5-10% of patients have a total colonic aganglionosis [8, 9]. HD is seen at approximately 5000 live births and is more common in boys. The genetic aspect of HD is still controversial. In the literature many cases with family history and also the association of HD with trisomy 21 and many other genetic diseases have been reported. In addition, new genetic mutations have been described in recent years [10, 11]. Clinical findings in HD, which is the most common organic cause of functional bowel obstruction seen in infancy vary according to the length of the affected intestine. In approximately 10% of newborns with long segment involvement HD, fever, abdominal distention, bile vomiting and diarrhea occur due to life-threatening enterocolitis. Infants who have short segment involvement HD have chronic constipation, abdominal distension, growth retardation. Unfortunately it is difficult to make a differential diagnosis from other causes of chronic constipation [12]. Although in healthy babies, passage of meconium is mostly observed in the first 24 hours, 95% of newborns with HD do not have passage of meconium in the first 24 hours of life [4, 7, 13]. It has been reported that babies with HD can remove meconium in the first 48 hour. The severity of the symptoms may vary from patient to patient, and in a group of patients, the late diagnosis is made after the newborn period, even at adulthood, due to the regression of the obstruction clinic as a result of enema. Late diagnosis of the disease may increase the risk of bacterial translocation, enterocolitis and colonic perforation, with high mortality [4, 14].

In this study, we aimed to retrospectively evaluate the features of the patients before making the diagnosis of HD.

Material and Method

The data of the patients who were diagnosed with Hirschsprung's disease (aged between 48 days and 40 months) between September 2016 and March 2018 in Faculty of Medicine, Department of Pediatrics were retrospectively analyzed. The study was approved by the Ethics Committee of the University with the decision dated 28.06.2018 and numbered 18.

In this study age, gender, percentile values of growth and development (height, weight, head circumference), neurodevelopment stages (ability to tonic neck reflex, ability to sit supported and unsupported ability to sit), first meconium time, the most common complaints and physical examination findings were examined.

In addition it was evaluated that these patients have the numbers of applications to the health facility, receive the misdiagnoses and the treatments, until the to diagnosis is made from initial of the complaints.

Results

The mean age of the 19 patients was 7.42 months (48 days-40 months). Eight patients were younger than 6 months. 7 patients were between 6 months and 1 year of age. 4 patients were older than 1 year.

Sixteen (84%) of the patients were male and 3 (16%) of the patients were female.

Growth and development values were detected in 8 of the patients in the range of 10-25, in 6 of the patients in the 3-10 percentile range and 5 of the patients were under the 3rd percentile.

The neurodevelopmental stages (tonic neck reflex, ability to sit supported, ability to sit unsupported) of all patients were similar with their peers of healthy.

The first meconium excretion time of the patients was an average of 3 days (49 hours-5 days).

It was determined that all of the patients had delay of meconium passage. And also the most common complaints of patients included abdominal distention in 19 (100%) patients, constipation in 15 (78.94%) patients, vomiting in 12 (63.15%) patients, fever in 7 (36.84%) patients, diarrhea in 4 (21.05%) patients and recurrent urinary tract infection in 3 patients.

Physical examination of patients determined abdominal distention in 19 (100%) patients, bile vomiting in 9 (47.36%) patients, fever in 5 (26.31%) patients, paleness in 15 (78.94%) patients, fecaloid mass in 7 (36.84%) patients and weakness in 11 (57.89%) patients. After the rectal examination explosive style stool-gas output was observed in all patients.

Four patients who have vomiting, abdominal distension, severe and malodorous diarrhea were in the clinical picture of enterocolitis. An intraoperative diagnosis was made in a patient with clinical picture of intestinal perforation (Table 1).

Complaints of the patients applying to the health facility included delayed meconium passage, constipation, abdominal distension, vomiting, weakness, restlessness, severe diarrhea (enterocolitis finding).

It was determined that the number of applications with these complaints to the health facility was an average of 4.3 times / 6 months.

It was determined that patients received misdiagnosis such as chronic constipation (7patients), infantile colic (4patients), recurrent urinary tract infection (3patients), lactose intolerance (3patients), gastro-esophageal reflux (2patients), metabolic disease (2 patients) and pyloric stenosis (1patient). And also it was determined that patients exposed to costly tests and unnecessary treatments such as advanced imaging methods and laxative medication, gas drop, antibiotic treatments, lactose free formula, antireflux therapy (Table 2).

Discussion

In developing countries, 90% of patients with HD are diagnosed within one year after birth. [11, 17]. This disease is more common in boys than girls. Complication rates of HD are higher

Table 1. Features of patients

	n	%
Demographic data		
Mean age	7.42	
<6 month	8	42.1
6month-1 year	7	36.8
>1 year	4	21
Gender		
Male	16	84.2
Female	3	15.8
Complaints		
Abdominal distansion	19	100
Constipation	15	78.9
Vomiting	12	63.1
Fever	7	36.8
Diarrhea	4	21
Urinary infection	3	15.8
Physical examination		
Abdominal distansion	19	100
Bile vomiting	9	44.3
Fever	5	26.3
Paleness	15	78.9
Fecaloid mass	7	36.8
Weakness	11	57.9
Explosıve Style Stool/Gas	19	100
Growthand Development Assessment		
>3 percentile	5	26.3
3-10 percentile	6	66.6
10-25 percentile	8	42.1

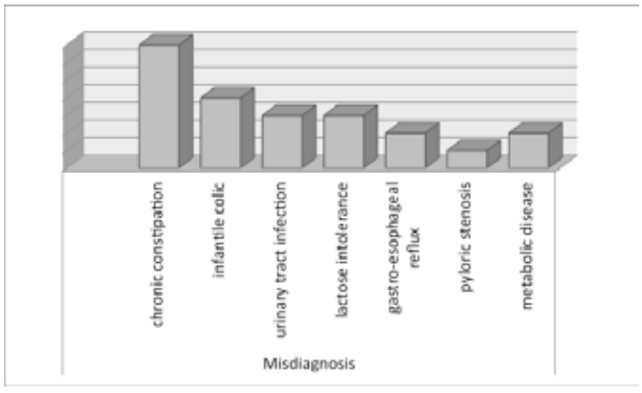

Table 2. Misdiagnosis of patients

in patients with the late diagnosis. Often, this disease may be presented with intestinal obstruction in newborns and with enterocolitis or chronic constipation in older children. The fact that if HD is not recognized early it causes an increase in intraluminal pressure in the dilated colon segment which is proximal to the aganglionic bowel segment. Unfortunately this state may cause decrease in intestinal blood flow, impaired mucosal barrier, excessive proliferation of bacteria, enterocolitis, perforation in the dilated colon segment, and leads to it has a higher risk of morbidity and mortality. Therfore it is important to recognize the disease before from the onset of enteroclolitis for the reduction of morbidity and mortality risk in HD. In our study the mean age of the 19 patients was 7.42 months (48 days-40 months). Eight patients were younger than 6 months, 7 patients were between 6 months and 1 year of age ve 4 patients were older than 1 year. Sixteen (%84) of the patients were male and 3 (%16) of the patients were female.

The most common clinical signs in older children are chronic constipation and malnutrition [4, 5, 11]. Growth and development values were detected in the patients in the range of 10-25, in 6 of the patients in the 3-10 percentile range and in 5 of the patients were found to be under the 3rd percentile. In addition it was determined the neurodevelopmental stages (tonic neck reflex, ability to sit supported, ability to sit unsupported) of all patients were similar with their peers of healthy.

In the 90% of babies with HD, meconium passage is not observed in the first 24 hours of their life. Normally, in the term newborn babies meconium passage is observed in the first 24 hours of their life while in preterm newborns meconium passage is observed in the first 48 hours of their life. If the meconium passage does not observed within the first 48 hours, HD should be considered [4, 6, 11]. In our study it was determined that all of the patients had a delay of meconium passage. And also the first meconium excretion time of the patients was an average of 3 days (49 hour-5 days).

Clinic course of HD may vary from intestinal obstruction in the newborn babies to chronic constipation in older children depending on the affected extent of the colon. In approximately 80% of patients reduced intestinal bowel movements, refuse feeding and progressive abdominal distention occur in the first few months of their life. Patients who were not early diagnosed in the infant period may present the clinical picture of enterocolitis. And also in some infants with normal passage of meconium present a clinical picture of chronic constipation [4, 5, 15]. These patients are prone to urinary tract infection because of the mass effect of fecaloma accumulated in the dilated bowel segment on the bladder. In this patients recurrent urinary tract infections, dilated bladder or even hydronephrosis may occur, due to urinary retention secondary to urinary system compression [2, 11]. The most common complaints in patients are delayed meconium passage, abdominal distension, feeding intolerance and bile vomiting [4]. In our study it was determined that all of the patients had anamnesis of delayed meconium passage and that especially after the use of suppositories drugs there was explosive style stool-gas output. And also the most common complaints of patients included abdominal distention in 19 patients, constipation in 15 patients, vomiting in 12 patients, fever in 7 patients, diarrhea in 4 patientsand recurrent urinary tract infection in 3 patients. Patients with HD who are breastfed during infancy have well clinical symptoms rather than patients who are fed fomula in infant period [4, 5].

In our study, in accordance with the literature constipation complaints of 6 patients fed with breast milk were less severe than complaints of 5 patients fed with formula and the growth and development parameters of these patients were also better. On physical examination in HD, there are distended abdomen, empty rectum and after rectal examinationexplosive style defecation [4, 6, 16]. .Differential diagnosis should be made in older children due to abdominal distension and other chronic constipation. Generally, in these patients, constipation start in the infant period and the response to medical treatments is

low and there is palpable mass in the left lower quadrant of the abdomen due to fecalomas. In the rectal examination, the anus is located normally, there are no feces in the rectum, but when the finger is back pulled, there is an explosive malodorous feces and gas outflow.There may be intermittent intestinal obstruction episodes accompanied by pain and fever [4, 5, 11].

Physical examination of the patients determined the abdominal distention in 19 patients, bile vomiting in 9 patients, fever in 5 patients, paleness in 15 patients, fecaloid mass in 7 patients and weakness in 11 patients. The anus of all patients was located in a normal position. After the rectal examination explosive style stool-gas output were observed in all patients. Four patients who have vomiting, abdominal distension, severe and smelly diarrhea were in the clinical picture of enterocolitis. An intraoperative diagnosis was made in a patient who has in the clinical picture of intestinal perforation.

In some infants, meconium passage is normal and the disease can be seen as chronic constipation. Constipation, defined as difficult defecation (less than 3 times per week and continued at least two weeks) is the most common gastroenterological complaint in the childhood, [13, 16-18]. Anamnesis and physical examination are the most important steps in the evaluation of the patient presenting with constipation [11, 19]. It should first be decided whether the disease is organic or functional [2, 4, 19].

Weight loss, growth retardation, bloody stool, constipation from early infant period; fever, accompanied by vomiting and other system findings should thought to have organic causes. Allergic diseases(celiac disease, lactose intolerance) as well as first defecation time in children with constipation should be questioned and also it must be learned whether Hirschsprung's disease take place in the family anamnesis of patients with constipation.

In our study, it was learned that 17 patients with ages ranging from 6 months to 40 months had complaints of difficulty defecation since the first days of their lives and did not respond to the treatment. It was determined that the number of applications to the health facility was an average 4.3 times / 6 months due to complaints of constipation however there was no response to the treatment (laxative and suppository drugs). And also it was determined that patients received misdiagnosis such as chronic constipation (7 patients), infantile colic (4 patients), recurrent urinary tract infection (3 patients), lactose intolerance (3 patients), gastro-esophageal reflux (2 patients), metabolic disease (2 patients) and pyloric stenosis (1 patient).

In this study it was determined that patients were exposed to costly tests and unnecessary treatments such as advanced imaging methods and laxative medication, gas drop, antibiotic treatments, lactose free formula, antireflux therapy (Table 2).

Consequently; although constipation can be mostly seen in all childhood age groups, the fact that it is not perceived as a serious condition by families and causes late diagnosis. However, the complaint of constipation may be a sign of serious organic disorders, especially in the newborn period [4, 13, 15, 21]. Therefore, family physicians and pediatricians should take into account the complaints of constipation and the child's habit of defecation and should question in detail and informed families to prevent late diagnosis. It should be kept in mind that organic causes such as HD should be considered in children who have history of delayed meconium passage in the anamnesis and who presented with complaints before the age of one year and whose growth retardation is detected in their examinations. In this way, the late diagnosis, complications, unnecessary and costly tests and treatments can be prevented in the HD.

The limitation of this study is that there is no evaluation of the characteristics of the patients in the postoperative period.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

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Conflict of interest

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